

DESCRIPTION *Rew*

RHEUMATOID ARTHRITIS GENE AND

METHOD FOR DIAGNOSING RHEUMATOID ARTHRITIS

5 This application is a 371 of PCT/JP00/01697 filed March 21, 2000.

① > Background of the Invention

Technical Field

② 71. Field of the Invention

The present invention relates to the disease gene of rheumatoid arthritis present in the human X chromosome and a method for diagnosing rheumatoid arthritis by detecting the presence of the disease gene or its expression product.

DESCRIPTION OF RELATED ART

Background Art

Although aspects, particularly the pathological process, of arthritis and arthritis mutilans which cause rheumatoid arthritis, have been clarified through various investigations, because most autoimmune diseases associated with rheumatoid arthritis developed or worsen into the disease only when various causative factors coincide, the interaction itself of multiple factors must be clarified to understand the disease and to develop appropriate methods of treatment.

The number of patients with rheumatoid arthritis in the world is 1% or less (N. Engl. J. Med. 322: 1277-1289, 1990), but among siblings of patients, over 8% develop the disease (Cell. 85: 311-318, 1996), which leads to the notion that some genetic factor may be involved. However, molecular genetic

CLAIMS

1. A disease gene for rheumatoid arthritis, which is a mutant of protooncogene Dbl transcribing an mRNA that encodes the cDNA of which the sequence from the 2679th to 2952nd bases is shown in SEQ ID NO: 1, which disease gene transcribes an mRNA encoding the cDNA of which the region from the 20th to 274th bases in SEQ ID NO: 1 is substituted with the sequence of SEQ ID NO: 2.
2. The cDNA of the disease gene of claim 1.
(Amended)
3. A DNA fragment, which is a part of the disease gene of claim 1 ~~or the cDNA of claim 2~~, and contains the base sequence of SEQ ID NO: 3.
4. A protein which is an expression product of the disease gene of claim 1, wherein the amino acid sequence of the C-terminal is that shown in SEQ ID NO: 2.
5. A peptide, which is a part of the protein of claim 4, and contains partial sequence of the amino acid sequence shown in SEQ ID NO: 2.
(Amended)
6. An antibody against the protein of claim 4 ~~or the peptide of claim 5~~.
(Amended)
7. A method for diagnosing rheumatoid arthritis, comprising the detection of the mRNA from the disease gene of claim 1 ~~or the protein of claim 4~~, in a biological specimen.
8. A method for functionally complementing Dbl deficiency.